



spastic paraplegia type 4

Spastic paraplegia type 4 is part of a group of genetic disorders known as hereditary spastic paraplegias. These disorders are characterized by progressive muscle stiffness (spasticity) and the development of paralysis of the lower limbs (paraplegia). Hereditary spastic paraplegias are divided into two types: pure and complex. The pure types involve only the lower limbs, whereas the complex types also involve the upper limbs (to a lesser degree) and the nervous system. Spastic paraplegia type 4 is a pure hereditary spastic paraplegia.

Like all hereditary spastic paraplegias, spastic paraplegia type 4 involves spasticity of the leg muscles and muscle weakness. People with this condition can also experience exaggerated reflexes (hyperreflexia), ankle spasms, high-arched feet (pes cavus), and reduced bladder control. Spastic paraplegia type 4 generally affects nerve and muscle function in the lower half of the body only.

Frequency

The prevalence of spastic paraplegia type 4 is estimated to be 2 to 6 in 100,000 people worldwide.

Genetic Changes

Mutations in the *SPAST* gene cause spastic paraplegia type 4. The *SPAST* gene provides instructions for producing a protein called spastin. Spastin is found throughout the body, particularly in certain nerve cells (neurons). The spastin protein plays a role in the function of microtubules, which are rigid, hollow fibers that make up the cell's structural framework (the cytoskeleton). Microtubules are also involved in transporting cell components and facilitating cell division. Spastin likely helps restrict microtubule length and disassemble microtubule structures when they are no longer needed. Mutations in spastin impair the microtubules' ability to transport cell components, especially in nerve cells; researchers believe this contributes to the major signs and symptoms of spastic paraplegia type 4.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

In most cases, an affected person inherits the mutation from one affected parent. The remaining cases may result from new mutations in the gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- spastic paraplegia 4
- SPG4

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Spastic paraplegia 4, autosomal dominant
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1866855/>

Other Diagnosis and Management Resources

- GeneReview: Hereditary Spastic Paraplegia Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1509>
- GeneReview: Spastic Paraplegia 4
<https://www.ncbi.nlm.nih.gov/books/NBK1160>
- Spastic Paraplegia Foundation, Inc.: Treatments and Therapies
<http://sp-foundation.org/understanding-pls-hsp/treatments.html>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Neurologic Diseases
<https://medlineplus.gov/neurologicdiseases.html>
- Health Topic: Neuromuscular Disorders
<https://medlineplus.gov/neuromusculardisorders.html>
- Health Topic: Paralysis
<https://medlineplus.gov/paralysis.html>

Genetic and Rare Diseases Information Center

- Hereditary spastic paraplegia
<https://rarediseases.info.nih.gov/diseases/6637/hereditary-spastic-paraplegia>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Hereditary Spastic Paraplegia
<https://www.ninds.nih.gov/Disorders/All-Disorders/Hereditary-spastic-paraplegia-Information-Page>

Educational Resources

- Disease InfoSearch: Spastic paraplegia 4
<http://www.diseaseinfosearch.org/Spastic+paraplegia+4/6695>
- MalaCards: spastic paraplegia 4
http://www.malacards.org/card/spastic_paraplegia_4
- Merck Manual of Medical Information, Second Home Edition
<http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/spinal-cord-disorders/hereditary-spastic-paraparesis>
- Orphanet: Hereditary spastic paraplegia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=685

Patient Support and Advocacy Resources

- National Ataxia Foundation
<http://www.ataxia.org/>
- National Organization for Rare Disorders (NORD): Hereditary Spastic Paraplegia
<https://rarediseases.org/rare-diseases/hereditary-spastic-paraplegia/>
- RareConnect
<https://www.rareconnect.org/en/community/hereditary-spastic-paraplegia>
- Spastic Paraplegia Foundation, Inc.: About Hereditary Spastic Paraplegia
<http://sp-foundation.org/understanding-pls-hsp/hsp.html>

GeneReviews

- Hereditary Spastic Paraplegia Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1509>
- Spastic Paraplegia 4
<https://www.ncbi.nlm.nih.gov/books/NBK1160>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22spastic+paraplegia+4%22+OR+%22Paraplegia%2C+Spastic%22+OR+%22Spastic+Paraplegia%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SPG4%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- SPASTIC PARAPLEGIA 4, AUTOSOMAL DOMINANT
<http://omim.org/entry/182601>

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Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735583/>
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